MPZ gene

myelin protein zero

Normal Function

The *MPZ* gene provides instructions for making a protein called myelin protein zero. It is the most abundant protein in myelin, a protective substance that covers nerves and promotes the efficient transmission of nerve impulses. Specialized cells called Schwann cells, which wrap around and insulate nerves, are the only cells that make myelin protein zero. Schwann cells are part of the peripheral nervous system which connects the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound. Myelin protein zero is required for the proper formation and maintenance of myelin. This protein is an adhesion molecule, which means it acts like molecular glue. It plays a role in tightly packing the myelin around nerve cells (myelin compaction).

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

Researchers have identified more than 120 *MPZ* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 1B. Many of these mutations change single protein building blocks (amino acids) in myelin protein zero. Other *MPZ* gene mutations lead to a protein that is missing one or more amino acids. The altered myelin protein zero probably cannot interact properly with other myelin components, which may disrupt the formation and maintenance of myelin. As a result, peripheral nerve cells cannot activate muscles used for movement or relay information from sensory cells back to the brain, leading to the weakness and sensory problems characteristic of Charcot-Marie-Tooth disease.

Some *MPZ* gene mutations cause a severe form of type 1B Charcot-Marie-Tooth disease. Symptoms begin during infancy or early childhood and include delayed development of motor skills such as walking. The early-onset forms of Charcot-Marie-Tooth disease are sometimes called Dejerine-Sottas syndrome, congenital hypomyelination, or Roussy-Levy syndrome. Researchers believe that the *MPZ* gene mutations that cause the severe form of the disorder probably disrupt the formation of myelin during early development.

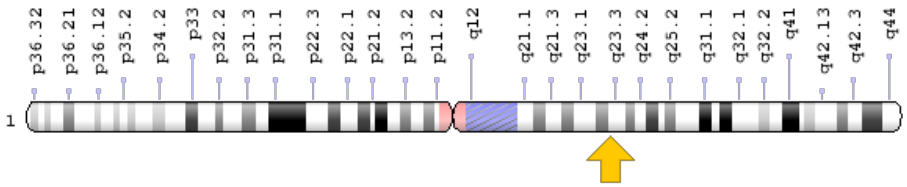
Several mutations in the *MPZ* gene cause other forms of Charcot-Marie-Tooth disease known as type 2I, type 2J, and dominant intermediate D. These forms of Charcot-Marie-Tooth disease, which often do not become evident until adulthood, affect the specialized outgrowths from nerve cells (axons) that transmit impulses to

muscles and other nerve cells. People with type 2J Charcot-Marie-Tooth disease may also have hearing loss and abnormalities in the opening of the eye through which light passes (the pupil). It is unclear how *MPZ* gene mutations cause these abnormalities.

Chromosomal Location

Cytogenetic Location: 1q23.3, which is the long (q) arm of chromosome 1 at position 23.3

Molecular Location: base pairs 161,303,593 to 161,309,972 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT1B
- HMSN1B
- MPP
- myelin glycoprotein P-zero
- myelin peripheral protein
- myelin protein zero (Charcot-Marie-Tooth neuropathy 1B)
- MYP0_HUMAN
- P0 Glycoprotein
- P0 Protein

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Cell Adhesion Molecules in Myelination
  https://www.ncbi.nlm.nih.gov/books/NBK28158/
- Basic Neurochemistry (sixth edition, 1999): The Myelin Sheath
  https://www.ncbi.nlm.nih.gov/books/NBK27954/

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1205
- Charcot-Marie-Tooth Neuropathy Type 2
  https://www.ncbi.nlm.nih.gov/books/NBK1285

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MPZ%5BTIAB%5D%29+OR+%28myelin+protein+zero%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- MYELIN PROTEIN ZERO
  http://omim.org/entry/159440

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MPZ%5Bgene%5D
- HGNC Gene Family: V-set domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/590
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7225
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=2
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4359
- UniProt
  http://www.uniprot.org/uniprot/P25189

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